TRPM4 gene

transient receptor potential cation channel subfamily M member 4

Normal Function

The *TRPM4* gene provides instructions for making a protein called transient receptor potential cation channel subfamily M member 4 (TRPM4). This protein acts as a channel, opening and closing at specific times to control the flow of positively charged atoms (cations) into and out of cells. The TRPM4 channel is embedded in the outer membrane of cells throughout the body, but it is abundant in heart (cardiac) cells and plays key roles in these cells' ability to generate and transmit electrical signals. TRPM4 channels play a major role in signaling the start of each heartbeat, coordinating the contractions of the upper and lower chambers of the heart, and maintaining a normal heart rhythm.

In addition to regulating electric signaling in the heart, the TRPM4 channel is important for the normal functioning of the immune system, the nervous system, the kidneys, and the pancreas.

Health Conditions Related to Genetic Changes

Brugada syndrome

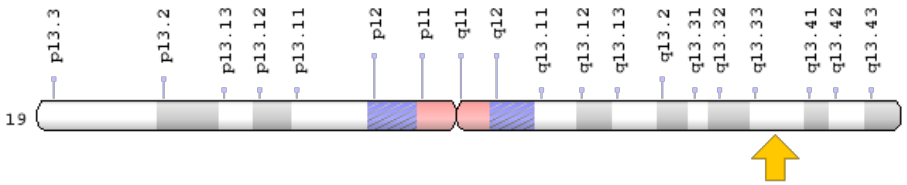
progressive familial heart block

A few mutations in the *TRPM4* gene have been found to cause progressive familial heart block. This condition alters the normal beating of the heart and can lead to fainting (syncope) or sudden cardiac arrest and death. The *TRPM4* gene mutations change single protein building blocks (amino acids) in the TRPM4 channel. The altered channels have increased activity at the cell membrane, which likely alters the cell's ability to generate electrical signals because of the increased cation flow. Cardiac cells with these altered channels have difficulty producing and transmitting electrical signals that coordinate normal heartbeats. Interruption of this signaling causes heart block. Death of these impaired cardiac cells over time can lead to a buildup of scar tissue (fibrosis), worsening the heart block.

Chromosomal Location

Cytogenetic Location: 19q13.33, which is the long (q) arm of chromosome 19 at position 13.33

Molecular Location: base pairs 49,157,741 to 49,211,841 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- calcium-activated non-selective cation channel 1
- hTRPM4
- long transient receptor potential channel 4
- LTrpC4
- melastatin-4
- transient receptor potential cation channel, subfamily M, member 4
- TRPM4_HUMAN
- TRPM4B

Additional Information & Resources

Educational Resources

- TRP Ion Channel Function in Sensory Transduction and Cellular Signaling Cascades (2007): The Ca2+-Activated TRP Channels: TRPM4 and TRPM5 https://www.ncbi.nlm.nih.gov/books/NBK5257/

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28TRPM4%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY M, MEMBER 4
  http://omim.org/entry/606936

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_TRPM4.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=TRPM4%5Bgene%5D
- HGNC Gene Family: Transient receptor potential cation channels
  http://www.genenames.org/cgi-bin/genefamilies/set/249
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17993
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/54795
- UniProt
  http://www.uniprot.org/uniprot/Q8TD43

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